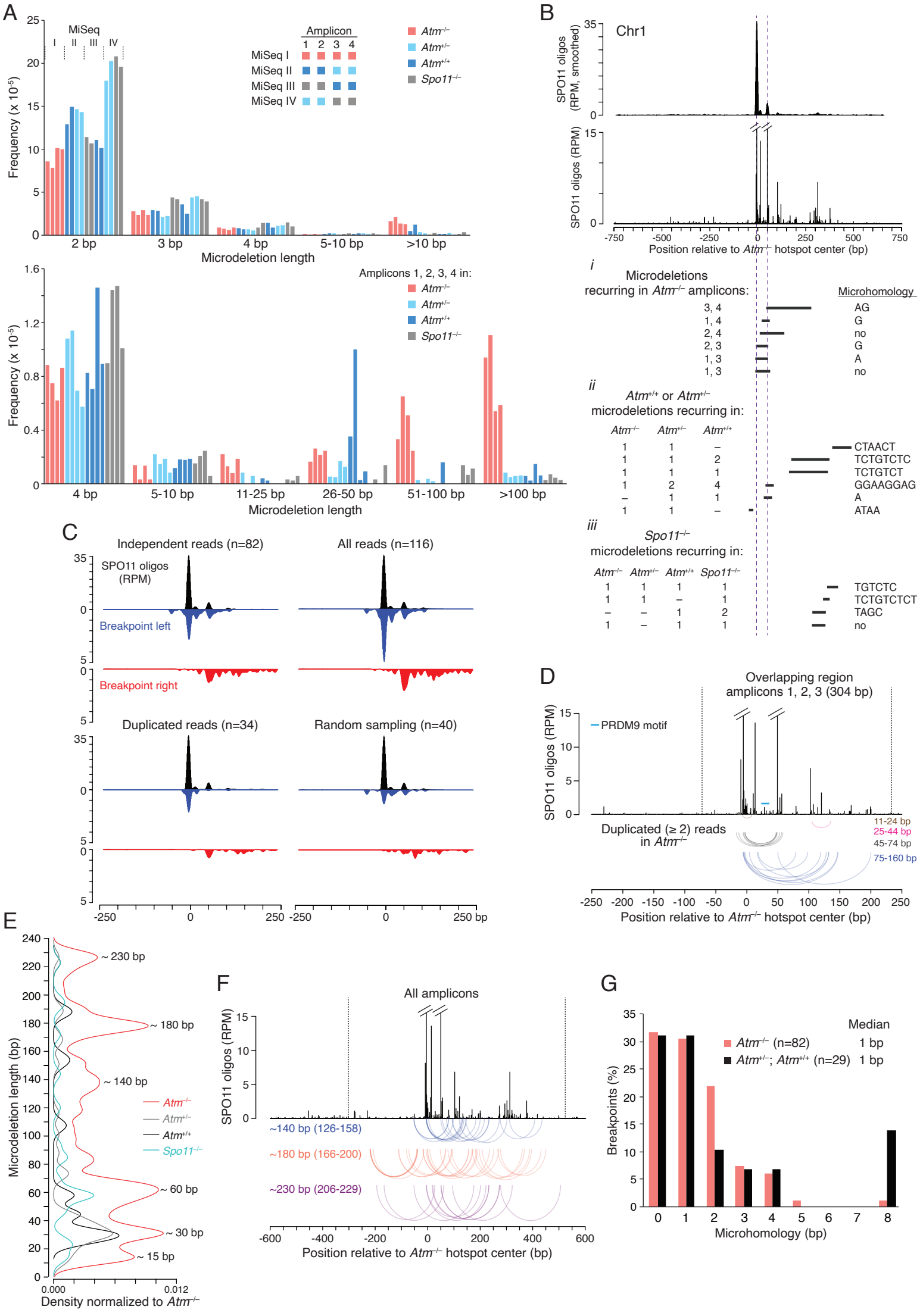
A
MiSeq
I
II
III
IV
Amplicon
1 2 3 4
MiSeq I
MiSeq II
MiSeq III
MiSeq IV
Atm−/−
Atm+/−
Atm+/+
Spo11−/−
Frequency (x 10-5)
2 bp
3 bp
4 bp
5-10 bp
>10 bp
Microdeletion length
Amplicons 1, 2, 3, 4 in:
4 bp
5-10 bp
11-25 bp
26-50 bp
51-100 bp
>100 bp
B
Chr1
SPO11 oligos (RPM, smoothed)
SPO11 oligos (RPM)
Position relative to Atm−/− hotspot center (bp)
i
Microdeletions recurring in Atm−/− amplicons:
Microhomology
3, 4 AG
1, 4 G
2, 4 no
2, 3 G
1, 3 A
1, 3 no
ii
Atm+/+ or Atm+/− microdeletions recurring in:
Atm−/− Atm+/− Atm+/+
1 1 –
1 1 2
1 1 1
1 2 4
– 1 1
1 1 –
CTAACT
TCTGTCTC
TCTGTCT
GGAAGGAG
A
ATAA
iii
Spo11−/− microdeletions recurring in:
Atm−/− Atm+/− Atm+/+ Spo11−/−
1 1 1 1
1 1 – 1
– – 1 2
1 – 1 1
TGTCTC
TCTGTCTCT
TAGC
no
C
Independent reads (n=82)
All reads (n=116)
SPO11 oligos (RPM)
Breakpoint left
Breakpoint right
Duplicated reads (n=34)
Random sampling (n=40)
bp
D
Overlapping region amplicons 1, 2, 3 (304 bp)
PRDM9 motif
SPO11 oligos (RPM)
Duplicated (≥ 2) reads in Atm−/−
11-24 bp
25-44 bp
45-74 bp
75-160 bp
Position relative to Atm−/− hotspot center (bp)
E
~ 230 bp
~ 180 bp
~ 140 bp
~ 60 bp
~ 30 bp
~ 15 bp
Microdeletion length (bp)
Density normalized to Atm−/−
F
All amplicons
SPO11 oligos (RPM)
~140 bp (126-158)
~180 bp (166-200)
~230 bp (206-229)
Position relative to Atm−/− hotspot center (bp)
G
Median
Atm−/− (n=82) 1 bp
Atm+/−; Atm+/+ (n=29) 1 bp
Breakpoints (%)
Microhomology (bp)